Experimental
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Clinical Article

Obstetrics; *Maternal Fetal Medicine and Perinatology*

The Analysis of the Fetal Abdominal Wall and Gastrointestinal Tract Abnormalities in a Single Tertiary Center

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ABSTRACT

OBJECTIVE: We aimed to evaluate the fetuses with gastrointestinal anomalies and anterior abdominal wall defects detected in the prenatal period in our clinic and to analyze their perinatal outcomes.

STUDY DESIGN: This retrospective study analyzed the data of 67 cases evaluated by the perinatology department of the Basaksehir Cam and Sakura City Hospital between January 2021 and May 2023 with a diagnosis of fetal anterior abdominal wall and gastrointestinal tract abnormalities.

RESULTS: A total of 67 cases were included in our study with a mean maternal age of 26.4 ± 3.1 years and a median gestational week of first assessment at our center of 22 weeks (12-39 weeks). The most common anterior abdominal wall malformations were omphalocele (n=29, 43.3%) and gastroschisis (6 cases, 9.0%), and the most common fetal gastrointestinal tract anomalies were duodenal atresia (n=10, 14.9%), dilated bowel (n=5, 7.4%), and intestinal atresia (n=4, 5.9%). While genetic testing was carried out in 18 cases (27%), chromosomal abnormality was found in 6 (9.0%) cases. Termination of pregnancy was performed in 6 cases (9.0%). Postnatally, surgery was performed in 49 cases (73.1%) with a diagnosis of postpartum omphalocele, duodenal atresia, gastroschisis, intestinal and anal atresia. An uneventful surgical course was recorded for 47 cases that underwent surgery.

CONCLUSION: The most common abdominal wall defects are omphalocele and gastroschisis, and the most frequent gastrointestinal anomalies are small bowel abnormalities (duodenal atresia, dilated bowel, intestinal atresia) in our study cohort. These abnormalities might be an isolated anomaly or part of a syndrome or associated with chromosomal abnormalities. The prognosis for infants with gastrointestinal malformation depends on the presence of associated anomalies or karyotype anomalies.

Keywords: Abdominal wall defects; Gastrointestinal tract abnormalities; Prenatal ultrasound

Gynecol Obstet Reprod Med (Article in Press)

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Submitted for Publication: 25.08.2024 Revised for Publication: 09.09.2024
Accepted for Publication: 22.10.2024 Online Published: 25.10.2024

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	DOI:10.21613/GORM.2023.1530

Introduction

Fetal anterior abdominal wall defects refer to a group of anomalies associated with defects in the anterior abdominal wall, the frequency is reported to be around 6 in 10,000 births (1). The most common defects are omphalocele and gastroschisis, and less commonly observed conditions include pentalogy of Cantrell, bladder and cloaca exstrophy, limb-body wall complex (LBWC), and OEIS (omphalocele, cloaca exstrophy, anus imperforatus, and spina bifida) complex (2,3). Prenatal diagnosis of omphalocele and gastroschisis is relatively straightforward, prenatal diagnosis rates of around 95-100% are reported in the fetal anatomy scan at 11-14 or the latest 18-22 weeks of gestation (4,5).

Fetal gastrointestinal tract anomalies are infrequent malformations with a global incidence of 3 to 6 cases every 1000 newborns. Abdominal cystic structures and intestinal atresia

How to cite this article: *Bolluk G. Oglak SC. Cetinkaya M. Bakirci IT. Demirali O. Cok M. Karakus HT. Onur Y. Yilmaz EZ. The Analysis of the Fetal Abdominal Wall and Gastrointestinal Tract Abnormalities in a Single Tertiary Center. Article in Press.*

or stenosis are the most common ones. The frequency of fetal intra-abdominal cystic lesions is reported to be approximately 1 in 1000 pregnancies (6,7). Intraabdominal cysts originate from organs in the fetal abdomen and pelvis. Most of them are of renal origin, but they may arise from the intestine, mesentery, ovary, pancreas, spleen, liver, and hepatic origin. Excluding those of renal origin, the most common cysts are ovarian cysts, gastrointestinal cystic duplications, liver and common bile duct cysts, meconium pseudocysts, and mesenteric and adrenal cysts (8).

Congenital gastrointestinal anomalies and anterior abdominal wall defects may result from genetic and environmental factors that are frequently encountered as fetal ultrasound (US) technology develops over time. While major and multiple anomalies may be life-threatening, minor anomalies may not require treatment. Prenatal diagnosis may change prenatal or postnatal management (9).

The primary diagnostic technique for these abnormalities is the US examination. When the US is insufficient, magnetic resonance imaging (MRI) can be utilized as a supplementary method (10). Intrauterine recognition of fetal gastrointestinal tract and abdominal wall anomalies and knowing their possible complications play a decisive role in pregnancy follow-up strategy, delivery method, and perinatal care (11).

In this study, we aimed to evaluate the fetuses with gastrointestinal anomalies and anterior abdominal wall defects detected in the prenatal period in our clinic and to analyze their perinatal outcomes.

Material and Method

The retrospective study was carried out at the Basaksehir Cam and Sakura City Hospital with the permission of Basaksehir Cam and Sakura City Hospital Clinical Researches Ethics Committee (approval date: 23.10.2023, number: 461). All procedures were carried out in accordance with the ethical rules and the principles of the Declaration of Helsinki.

This study was conducted by evaluating 67 cases referred from the external center or diagnosed for the first time with fetal gastrointestinal tract and abdominal wall anomalies between January 2021 and July 2023 in the Perinatology Department of Cam and Sakura City Hospital. Our hospital serves as a tertiary referral center in the region. Based on the differential diagnosis, thoracic anomalies, including congenital diaphragmatic hernia and esophageal atresia, were excluded from this study. Cases with incomplete medical records and lacking postnatal confirmation of the fetal abnormality were also excluded.

Fetal ultrasonographic (US) examination and diagnosis of these abnormalities were made by maternal-fetal medicine specialists using ultrasonic equipment with the option of color and a pulsed Doppler US with Arietta 850 US device (HITACHI, Japan). After the diagnosis of fetal anomaly, all cases were re-evaluated by the multidisciplinary committee, which consists of medical specialists involved in perinatal care, including maternal-fetal medicine specialists, obstetricians, neonatologists, pediatric surgeons, pediatric intensive care specialists, and clinical geneticists. Following this, information was given to parents about the significance of the diagnosis, whether surgical or non-surgical treatment modalities would be required after birth, the expected short and long-term prognosis of these cases, and information about the possibility of termination.

Maternal age, gestational week at diagnosis, reasons for referral to our clinic, presence of chromosomal anomalies, and prenatal-postnatal treatments were recorded.

The confirmation of prenatal diagnosis was made by postnatal follow-up in the first months of life, and/or by autopsy in cases of termination of pregnancy.

Statistical Analysis

IBM SPSS Statistics for Windows, Version 26.0 (Chicago, IL, USA) was used for data analysis. The Shapiro-Wilk test was used to assess the normality of the data.. Continuous variables that meet the normal distribution assumption were presented as mean±standard deviation and the remaining variables were presented as median (minimum-maximum). Descriptive statistics were presented as frequencies and percentages for categorical variables.

Results

A total of 1244 pregnant women who had major fetal abnormalities were assessed in our center during the study period. Over two years, 67 fetuses were diagnosed at our center as having fetal gastrointestinal tract and abdominal wall anomalies. The antenatal prevalence of fetal gastrointestinal tract and abdominal wall anomalies in our perinatology unit was 5.4% among all cases of abnormality cases.

Demographic and clinical characteristics of the cases are shown in Table I. The median gestational age at first evaluation at our center was 22 weeks (ranging from 12 to 39 weeks), and the mean maternal age was 26.4±3.1 years. All pregnancies included in this study ended up in our hospital.

Table I: *Demographic and clinical characteristics of the cases*

Maternal age, years (mean±std)	26.4±3.1
Gravida, median (min-max)	2 (1-8)
Gestational week at diagnosis, median (min-max)	22 (12-39)
Chromosomal anomaly, n (%)	6 (9.0%)
Termination of pregnancy, n (%)	6 (9.0%)
Intrauterin fetal death, n (%)	3 (4.5%)
Gestational week at birth, median (min-max)	34 (12-41)
Birth weight, g, median (min-max)	2550 (?-3340)

Table II shows the classification of the fetal gastrointestinal tract and abdominal wall anomalies based on the prenatal and postnatal diagnosis of the cases in our study cohort. Among 67 cases, 56 cases (83.6%) had isolated fetal gastrointestinal tract or abdominal wall anomalies, while 11 cases (16.4%) had additional congenital abnormalities. A total of 12 different types of fetal gastrointestinal tract and abdominal wall anomalies were identified prenatally. The following list shows the frequency of these anomalies: omphalocele (n=29, 43.3%, Figure 1), duodenal atresia (n=10, 14.9%), gastroschisis (n=6, 9.0%, Figure 2), dilated bowel (n=5, 7.4%), intestinal atresia (n=4, 5.9%), gall bladder atresia (n=3, 4.5%), anal atresia (n=3, 4.5%), intestinal cystic lesions (n=3, 4.5%), Pentalogy of Cantrell (n=1, 1.5%), OEIS Complex (n=1, 1.5%), gall bladder duplication (n=1, 1.5%), and hepatomegaly (n=1, 1.5%).

Table II: *Classification of fetal gastrointestinal tract and abdominal wall anomalies*

Diagnosis	n	%
Omphalocele, n (%)	29	43.3%
Duodenal atresia, n (%)	10	14.9%
Gastroschisis, n (%)	6	9.0%
Dilated bowel, n (%)	5	7.4%
Intestinal atresia, n (%)	4	5.9%
Gall bladder atresia, n (%)	3	4.5%
Anal atresia, n (%)	3	4.5%
Intestinal cystic lesions, n (%)	3	4.5%
Pentalogy of Cantrell	1	1.5%
OEIS Complex	1	1.5%
Gall bladder duplication, n (%)	1	1.5%
Hepatomegaly, n (%)	1	1.5%
Total	67	100%

Chromosomal analysis was performed in 18 cases (27%). However, chromosomal abnormality was found in 6 cases (9.0%). Of these, two fetuses had trisomy 21 (both cases had duodenal atresia), one had trisomy 18 (omphalocele), one had trisomy 13 (omphalocele), one had trisomy 15 (jejunal atresia), and one had t(12;14) (p13.3;q2) translocation (duodenal atresia).

Parents opted for termination of pregnancy (TOP) in 9 cases (13.5%); of which, 6 fetuses had chromosomal anomalies, one had Cantrell of Pentalogy, one had OEIS complex, and one had omphalocele with hydrops fetalis. Of the 6 pregnancies that were terminated, 2 fetuses had trisomy 21, one had trisomy 18, one had trisomy 13, one had Cantrell of Pentalogy, and one had omphalocele with hydrops fetalis. In total, intrauterine fetal death was observed in 3 cases (4.5%). Of these, one fetus had duodenal atresia, one had omphalocele, and one had OEIS complex. No cases received intrauterine treatment during the antenatal follow-up period. All live births were survived and neonatal death was not observed in any case.

In our study, MRI was not required for the diagnosis of any cases. Prenatally, no procedure was performed for diagnoses and treatment. Postnatally, surgery was performed in 49 cases (73.1%) with a diagnosis of postpartum omphalocele, duodenal atresia, gastroschisis, intestinal and anal atresia. An uneventful surgical course was recorded for 47 cases that underwent surgery.

Discussion

The embryological development of the fetal abdominal wall starts during the third week of pregnancy. During this period, lateral body folds form from the mesoderm and ectoderm, gradually closing around the midline to enclose the abdominal organs. By the sixth week, the intestines temporarily herniate into the umbilical cord as they grow rapidly. This physiological herniation resolves by the 10th week, with the intestines returning to the abdominal cavity. The abdominal muscles, fascia, and skin layers continue to develop throughout the first trimester, completing the formation of the abdominal wall (12). The closure of the ventral body wall can be dis-

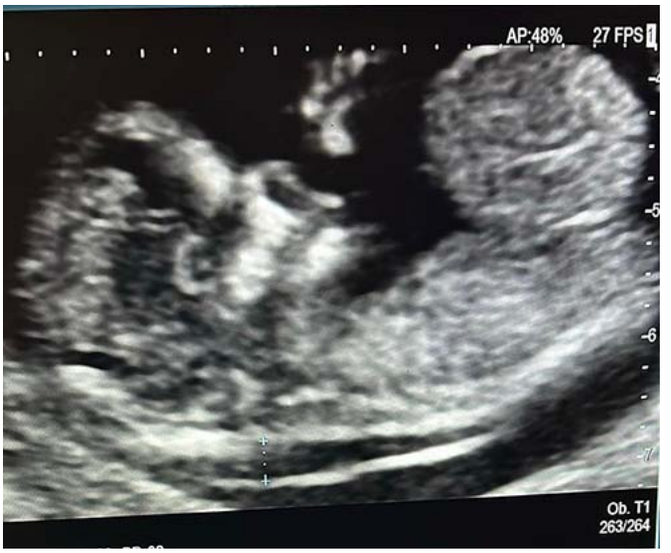

Figure 1: *A case with omphalocele*

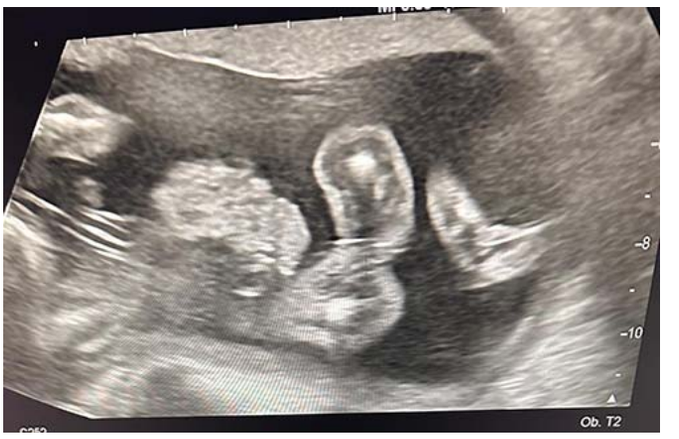

Figure 2: *A case with gastroschisis*

rupted between the third and fourth weeks of development if the lateral body folds fail to fuse properly at the midline. However, for gastroschisis to develop, this disruption must occur between the eighth and eleventh weeks. By this time, the midgut has already herniated, and the umbilical cord, particularly its vascular structures, is fully formed, making this period critical for the occurrence of the defect (12,13).

Prenatal US can identify most of the major anomalies in the fetal gastrointestinal tract. However, these anomalies are challenging to evaluate because the abdomen involves numerous organs, the bowel's anatomy is intricate, and US findings are variable. Moreover, the normal appearance of the fetal bowel changes at different stages of gestation, adding to the complexity (14). Based on the American Institute of Ultrasound in Medicine and the American College of Radiology guidelines, every fetal US examination should include documentation of five anatomical features: the abdominal circumference, the fluid-filled stomach, the cord insertion and adjacent abdominal wall, the kidneys and surrounding structures, and the urinary bladder (15). By routinely evaluating all five anatomical features, most of the fetal abdominal abnormalities can be identified. A recent study showed that 96% of fetal anomalies were accurately detected when only these five specified views were reviewed (16).

Early diagnosis is crucial for fetal gastrointestinal anomalies, just as it is for any other fetal structural abnormality. Prenatal detection and a better understanding of the developmental process of these anomalies enable earlier intervention and proper prenatal counseling for parents about outcomes and possible complications. It is also essential to determine whether the specific defect is an isolated anomaly or part of a syndrome. As known, the US is a noninvasive, cost-effective, and widely accessible method for evaluating fetal anatomical structures. Therefore, the US is commonly used to screen for fetal anomalies and can provide real-time imaging without ionizing radiation. Most US scans effectively diagnose these abnormalities (17,18). This study analyses fetal gastrointestinal and abdominal wall anomalies diagnosed during the prenatal period. The median age at the time of diagnosis in our study cohort is 22 weeks, though some cases can be identified as early as the first trimester. This underscores the critical importance of comprehensive US education for healthcare professionals. Expertise in the US allows for the early detection of anomalies and conditions that may otherwise go unnoticed until the later stages of gestation. Enhanced US training equips clinicians with the skills necessary to perform precise and accurate assessments, ultimately improving outcomes through early diagnosis and timely intervention.

According to the European registries included in the Eurocat network in 2011, the total prevalence of gastroschisis was reported as 3.09 per 10,000 births, with a live birth prevalence of 2.63 per 10,000 births. The corresponding figures for omphalocele were 3.29 and 1.13 per 10,000 births (19). Barisic et al. reported that omphalocele and gastroschisis are the most prevalent congenital abdominal wall anomalies in their study, and 19 congenital malformation registries were collected from 11 European countries (20). Omphalocele is characterized by a defect in the midline anterior abdominal wall, in which the abdominal organs protrude into the base of the umbilical cord, enclosed by a membranous sac. Gastroschisis is typically a small defect in the anterior abdominal wall, frequently located to the right of the umbilical ring, resulting in herniation of the abdominal contents into the amniotic cavity without a surrounding membrane (21). Both abnormalities are commonly identified prenatally through US examination. Likewise, the two most common abdominal wall defects are omphalocele (43.3%) and gastroschisis (9.0%) in our study cohort. The prenatal identification of associated malformations and chromosomal abnormalities is considered to affect perinatal outcomes. Consequently, a thorough and prompt examination of other structural malformations and karyotyping should be conducted whenever an abdominal wall defect is detected (22). The differential diagnosis of omphalocele and gastroschisis is crucial since fetuses with omphalocele have an increased risk for chromosomal abnormalities. In the literature, chromosomal abnormalities have been observed in approximately 30% of fetuses with omphalocele. Also, omphalocele can be associated with several syndromes, including Beckwith-Wiedemann syndrome, Charge syndrome, Meckel-Gruber syndrome, and so on. The prognosis largely depends on the associated congenital anomalies and syndromes. Therefore, prenatal diagnosis of this defect should alert the clinician to the probability of omphalocele-related disorders and familial inheritance and prompt thorough genetic counseling for these abnormalities (23). In our study, we found that two fetuses with omphalocele had chromosomal abnormalities; of these, one fetus had trisomy 18, and one had trisomy 13. Both of these pregnancies experienced TOP following the karyotype analysis. Since karyotype analysis was not performed in all pregnancies with omphalocele, we cannot accurately determine the exact rate of chromosomal anomalies in our study group.

A broad range of congenital anomalies can impact the gastrointestinal tract, with some becoming evident shortly after birth and others not appearing until later in childhood or adulthood. The incidence of gastrointestinal anomalies varied from 3.2% to 29% as noted by various authors (24). Likewise, the incidence of fetal gastrointestinal tract and abdominal wall anomalies in our maternal-fetal medicine unit was 5.4% among all fetal anomaly cases. Also, similarly to the literature (25), the most common fetal gastrointestinal tract abnormalities are small bowel abnormalities (duodenal atresia, dilated bowel, intestinal atresia) in our study. Prenatal diagnosis of bowel abnormalities is challenging due to the bowel's variable appearance throughout gestation and the varied manifestations of the same pathological condition (25). The possibility of

gastrointestinal anomalies being associated with chromosomal abnormalities should be taken into account. Aneuploidies, including trisomy 21, 13, and 18 may be linked to bowel malformations (26). In our cohort, two fetuses with duodenal atresia had trisomy 21, one fetus with duodenal atresia had t(12;14) (p13.3;q2) translocation, and one fetus with jejunal atresia had trisomy 15.

Surgical intervention after birth was needed in 49 cases (73.1%) among those with postnatal diagnosis. In our study, none of the cases underwent surgical treatment during the intrauterine period. Additionally, Cass et al. recently reported that fetal abdominal cysts do not commonly benefit from fetal intervention. However, rare conditions, including very large ovarian or hepatic cysts that cause compression of adjacent organs and bowel obstruction which can result in polyhydramnios create an exception (27).

The primary focus in managing a neonate with a cystic abdominal lesion is establishing an accurate diagnosis and ruling out any related conditions. A physical examination should assess for any dysmorphic features, with a referral to a genetics specialist if necessary. The timing of further evaluation depends on symptoms, such as prenatal bowel dilation or pressure on surrounding structures, as well as the suspected diagnosis. Initial imaging typically involves plain X-rays and the abdominal US. Additional studies, such as CT scans or MRI, may be used, especially for liver or pancreatic lesions. Surgical removal is often recommended for these lesions, although the timing may vary based on individual circumstances (27).

This study highlights the importance of prenatal diagnosis in a comprehensive perinatal approach to the congenital gastrointestinal tract and abdominal wall lesions. As known, informing the obstetrician and alerting the pediatric surgical team for resuscitation, immediate postnatal evaluation, and timely intervention can reduce neonatal mortality and improve long-term infant survival. Postnatal treatment modalities and thus, prognosis of the infant have improved over the years. Deliveries of these cases should be planned in tertiary centers where necessary intervention and neonatal care can be provided by a multidisciplinary team. However, there may be some possible limitations in this study. The main limitation is the retrospective design of this research and the relatively low sample size. Other limitations include karyotype analysis was not performed in all patients and the absence of long-term postnatal outcomes. Another significant limitation of our study is the inability to confirm diagnoses through autopsy in cases where pregnancy termination or intrauterine fetal demise occurred.

Conclusion

The most common abdominal wall defects are omphalocele and gastroschisis, and the most frequent gastrointestinal anomalies are small bowel abnormalities (duodenal atresia, dilated bowel, intestinal atresia) in our study cohort. These abnormalities might be an isolated anomaly or part of a syndrome or associated with chromosomal abnormalities. The prognosis for infants with gastrointestinal malformation depends on the presence of associated anomalies or karyotype anomalies.

Declarations

Ethics approval and consent to participate: All participants signed informed written consent for using data before being enrolled in the study. The study was reviewed and approved by the Basaksehir Cam and Sakura City Hospital Clinical Researches Ethics Committee (approval date: 23.10.2023, number: 461). All procedures were performed according to the Declaration of Helsinki.

Availability of data and materials: The data supporting this study is available through the corresponding author upon reasonable request.

Competing interests: The authors declare that they have no competing interests.

Funding: This study didn't receive any financial support from any organization, institution, or funding agency.

Acknowledgment: We are grateful to all participants and their families who spent precious time participating in this research program. We are also thankful for the tireless efforts of the research team members.

Authors' contributions: Concept: GB, SCO, MC, ITB, EZY; Design: GB, SCO, ITB, OD, MC, HTK, YO; Data Collection or Processing: GB, MC, ITB, MC, HTK, YO; Analysis or Interpretation: GB, SCO, ITB; Literature Review: GB, SCO, EZY; Writing: GB, SCO; Critical Review: SCO.

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